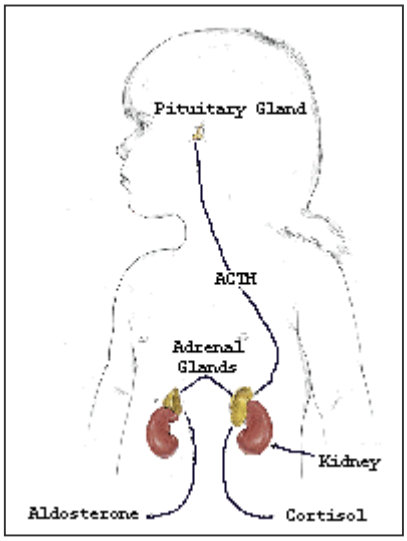
Congenital Adrenal Hyperplasia (CAH)
General Overview

Q. What is CAH?

A. It is a condition in which the adrenal glands do not produce normal amounts of certain essential hormones. Hormones are the chemicals produced by glands that regulate the function of other organs. Treatment is required for CAH in order to prevent serious illness and problems with growth and development.

Q. Where are the adrenal glands and what do they do?

A. The adrenal glands are two walnut-sized glands located on top of the kidneys. They produce many different hormones which enable the body to function normally. The three main groups of hormones which could be involved in CAH are:



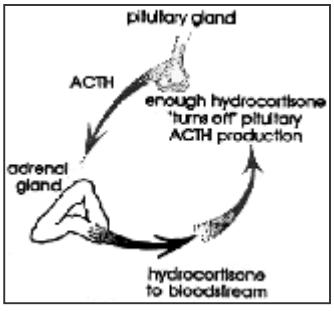
Glucocorticoids - (specifically cortisol), affects how the body uses protein, carbohydrate and fat to maintain the process of growth and the response to injury and infections.

Mineralcorticoids - (specifically aldosterone), affects how the kidneys regulate water and salt in the body.

Androgens or Male Hormones - affects body growth and the development of secondary sex characteristics.

Q. How do the adrenal glands normally produce hormones?

A. The process of producing the above noted hormones begins in the brain. It is controlled by the pituitary, or "master gland", a pea-sized gland located at the base of the brain. As the illustration shows, the control of hormones is a "thermostat-like" mechanism. Cortisol is the adrenal hormone which serves as a signal to the pituitary of the adrenal hormone levels in the body. When the amount of cortisol is too low, the pituitary senses it and releases a hormone, ACTH. The ACTH then stimulates the adrenal glands to produce more cortisol. When enough cortisol has been produced to meet the body's needs, no extra stimulation is needed, and the ACTH level drops to its usual level. This in turn slows the production of the other adrenal hormones.



Q. What causes the adrenal glands not to function properly in children with CAH?

A. In order for the adrenal glands to properly regulate the hormones they produce, certain protein substances called enzymes are required. Children with CAH have an inborn shortage of one of these enzymes. Without the enzyme, the adrenal glands can never make enough cortisol to meet the body's needs, therefore, the "thermostat" is not able to function properly. This results in an underproduction of cortisol and aldosterone, and overproduction of androgens.

Q. What are the effects of having CAH?

A. The abnormal production of adrenal hormones leads to the following consequences if untreated:

1. Too little cortisol- makes the body unable to respond to physical stress.
2. Too little aldosterone- results in the potentially life-threatening problem of excessive salt loss.
3. Too much androgens leads to:
    a.) abnormally rapid growth
    b.) early appearance of underarm, facial and body hair.

Q. Is there only one form of CAH?

A. There are several forms of CAH, but the 21-hydroxylase enzyme defect is by far the most common and accounts for over 90% of the cases diagnosed. The other forms are due to different enzyme defects in the production of the adrenal hormones and result in some different medical problems.

Q. What are the chances that a child will be born with CAH?

A. About one in every 15,000 babies is born with CAH. It is a genetic disorder in which the child inherits one non-working gene from each parent. Therefore, once you have a child with CAH, there is a 25% (1 in 4) chance with each pregnancy of having another affected child. There is testing available to identify if a child is affected before birth (prenatal diagnosis).

Q. What is the treatment for CAH?

A. The treatment for CAH is steroid replacement therapy to achieve normal hormone levels to allow for regular development.

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